

# A Five Years Study of Occurrence and Associated Risk Factors for Birth Defects in a Tertiary Care Hospital in Central Nepal

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## ABSTRACT

**Introduction:** Congenital anomalies account for 7.0% of neonatal deaths in Nepal. The present study was carried out to determine the overall rate of congenital malformations, incidence and prevalence in live births, still birth and incidence affecting various organ systems, at Bharatpur Hospital, Nepal.

**Methods:** All the intramural deliveries between Jan 2015 to Dec 2019 were included in the study. All the newborns were looked for congenital malformations after birth within seven days. Antenatal ultrasonography findings were noted. 2D echocardiography was also used for all congenital heart diseases, along with routine X-ray chest. A total of 131 babies with congenital problem were studied and the information was recorded in WHO NBBB Proforma. Data were recorded in MS Excel and SPSS 16 version was used for analysis.

**Results:** Out of the total 60160 deliveries, 131 (0.21% of total birth) were with congenital malformations, sex wise distribution was 65 (49.5%) females and 63 (48.7%) males and three (1.8%) were ambiguous. Oro-facial malformation (49, 37.4%) was the commonest form of malformation followed by the musculoskeletal system (31, 23.6 %), centre nervous system (31, 23.6%) and congenital malformations of genital organs (8, 6.0%).

**Conclusions:** The incidence of congenital malformation in this study was 0.21%. Females were more common than males and oro-facial malformation was the commonest type of malformation. Lack of antenatal visit, lack of folic acid during pre-conception period and low socioeconomic status were the commonest risk factors.

**Key words:** birth defect; cleft palate; live birth



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## INTRODUCTION

Birth defects are defined as abnormalities of structure, function or metabolism that are present at birth and result in physical, mental disability or mortality.<sup>1</sup> These defects may be isolated or multiple and are due to multiple etiological factors.<sup>2,3</sup> The prevalence and types of congenital anomalies varies from place to place and population to population.<sup>4-7</sup> The birth prevalence of congenital anomalies in the United States is 2-3%, United Kingdom is 2% and in South Africa it is 1.49%.<sup>8-11</sup>

The known causes of birth defects are mostly genetic effect modified by environmental factors which may be prevented. We found limited studies regarding the prevalence and risk factors about congenital birth defects in Nepal so we conducted a study to determine the prevalence and association of certain risk factors for birth defects occurring in newborn in our institute in central Nepal.

## METHODS

This is a hospital based cross sectional study conducted in a tertiary level hospital located in Chitwan, Nepal after taking ethical approval from Institutional review committee. We evaluated all the deliveries from 1 Jan 2015 to 31 Dec 2019. We analysed all the babies born in the hospital with one of the birth defects diagnosed at birth and until seventh day, both live births and stillbirths, and gestational age ( $\geq 22$  weeks or  $\geq 500$  gram).

All the newborns were looked for congenital malformations soon after birth and everyday during routine ward rounds. Maternal age, gestational age, sex, community, birth weight, birth order and consanguinity were documented. Maternal illness, ingestion of drugs, exposure to radiation and complications of labor was recorded. Antenatal ultra sonogram (USG) findings were noted. Relevant radiological, haematological and genetic tests were carried out. A meticulous general and systemic examination was carried out by a consultant and on duty doctors at the time of birth to detect any malformations. Ultrasound was employed routinely to detect multiple congenital anomalies and to rule out a majority of the internal congenital anomalies. 2D echocardiography screening was also done for all suspected

congenital heart diseases along with routine X-ray chest. The major malformations were divided into oro-facial, central nervous system (CNS), musculoskeletal, gastrointestinal, genitourinary, cardiovascular system (CVS), syndromes and miscellaneous disorders. Chromosomal abnormalities like Down's syndrome was diagnosed on the basis of clinical findings only.

Genetic and newborn metabolic assessments were not performed due to the lack of resources and facilities. After diagnosing congenital anomaly, informed consent was taken from parents and structured WHO-SEARO designed Birth Defect Proforma was filled. The pattern of congenital anomalies along with their system wise distribution was documented. The risk factors associated with malformations and their outcomes were also documented. The parents were interviewed regarding detailed maternal and antenatal history including maternal and paternal age, period of gestation, gravida, ethnicity, religion, socioeconomic status and number of antenatal care visit. For gestational maturity, the Nationwide Inpatient Sample coding system defined by preterm birth and term birth as delivery before and after 37 completed weeks of gestation respectively was used. Gravida was divided into two categories that is primi being first time pregnant and multi being pregnant for two or more times.

Pre-conception risk factors, family history of mother and father, first and second degree relatives and previous child with congenital anomalies were also recorded into the study. A marriage has been considered consanguineous when that is found to have occurred between a male and a female who are blood-related, e.g. between brother and sister, between first cousins. Data was entered in MS Excel and analysed on SPSS 16.

## RESULTS

Within the study period of five years, total delivery in the hospital were 60,160 out of which 131 babies were born with congenital anomalies, making the prevalence of 2.1 per 1000 live births and incidence of 0.21%. Among 131 neonates with congenital anomalies, 63 (48.7%) were males and 65 (49.5%) were females and three (1.8%) had disordered sexual development. The most frequent anomalies were oro- facial 49 (37.4%) followed by

**Table 1.** Distribution of the birth defects among the study population

System	Disordered sexual development n (%)	Female n (%)	Male n (%)	Total
Nervous system	0 (0)	16 (17.5)	15 (22.6)	31 (23.6)
Eye ear nose	0 (0)	3 (4.8)	2 (3.2)	5 (3.9)
Circulatory system	0 (0)	1 (1.6)	0 (0)	1 (0.7)
Respiratory system	0 (0)	1 (1.6)	0 (0)	1 (0.7)
Cleft lip and palate	0 (0)	23 (36.5)	26 (41.9)	49 (37.4)
Digestive system	0 (0)	1 (1.6)	2 (3.2)	3 (2.3)
Genital organs	3 (100)	1 (1.6)	4 (6.4)	8 (6.2)
Musculo-skeletal system	0 (0)	18 (24.4)	13 (20.7)	31 (23.6)
Down's syndrome	0 (0)	1 (1.6)	1 (1.6)	2 (1.5)
Total	3	65	63	128

musculoskeletal system 31 (23.6%), centre nervous system 31 (23.6%), genito-urinary eight (6%) and disordered sexual development in three (2.29%) (Table 1).

Among the birth defects the commonest is different form of cleft lip and cleft palate followed by talipes equino varus, different type of spins bifida and hydrocephalus, gastroschiasis ad anencephaly. The system wise involvement among 131 birth defect neonate showed the oro-facial defect (49, 37.4%) was the commonest, followed by musculoskeletal, CNS and genito-urinary (Table 2).

## DISCUSSION

Congenital anomalies if overt can be picked up easily at birth by trained paediatricians. Anomalies like congenital defects of the heart are apparent in four to seven days even if not apparent at or soon after birth. Sometimes patients are informed beforehand about the anomalies on antenatal ultrasounds, most common of these include

hydrocephalus, renal anomalies, heart defects, and anomalies of the lungs so that antenatal counselling can be done and necessary management plans can be laid out. The pattern and prevalence of congenital anomalies may vary over time or with geographical location. It also depends upon the environmental and genetic factors including socio-cultural, racial and ethnic variables.<sup>13</sup> With improved control of infections and nutritional deficiency diseases, congenital malformations have become important causes of perinatal mortality in developing countries.<sup>14</sup>

The joint World Health Organisation (WHO) and March of Dimes (MOD) meeting reported that 7% of all neonatal mortality and 3.3 million under five deaths were due to congenital anomalies.<sup>15</sup> Several studies have been done to determine the prevalence of birth defect and prevalence varied widely from 0.5% to 6.8%.<sup>16</sup>

Our study showed the prevalence of congenital anomalies to be 2.1 per 1000 live births and the incidence of congenital malformations in neonates to be 0.21% among live births which is lower than previous two studies done in maternity hospital (0.36%) and Western regional hospital (0.42%) in Nepal.<sup>17,18</sup> In England and United States of America, the prevalence is 2% and 2-3% respectively.<sup>5,18</sup> The prevalence of congenital malformation in our study is also comparable to another study done in United Arab Emirates (1%) and China (1.1%).<sup>19</sup>

In various studies done in developing countries like India<sup>19</sup> and Pakistan,<sup>20</sup> highest frequency of CNS anomalies and lowest frequency of congenital heart disease were demonstrated. The variation in the frequencies of different involved systems could be due to genetic background, geographical area, socioeconomic and nutritional status along with folic acid intake. Presently, more prevalence of cardiovascular anomalies may have been shown due to routine performance of echocardiography in all babies, as a part of antenatal screening.

In our study oro-facial malformation was the commonest birth defect followed by musculoskeletal system and centre nervous system resembling those found in other national studies and in developed countries like the United States of America and Europe.<sup>21</sup> Another study done in

**Table 2.** System wise birth defect in study population

System	Types of Anomalies	Number	%
<b>Nervous System</b> (n=31)	Anencephaly	5	16.1
	Congenital hydrocephalus	4	13.0
	Congenital hydrocephalus, unspecified	6	19.3
	Cervical spina bifida with hydrocephalus	2	6.4
	Lumbar spina bifida with hydrocephalus	7	22.5
	Sacral spina bifida with hydrocephalus	2	6.4
	Thoracic spina bifida without hydrocephalus	1	3.2
	Lumbar spina bifida without hydrocephalus	2	6.4
	Sacral spina bifida without hydrocephalus	2	6.4
<b>Eye, Ear, Nose</b> (n=5)	Other congenital malformations of eyelid	1	20.0
	Microphthalmos	1	20.0
	Congenital cataract	1	20.0
	Misplaced ear	1	20.0
	Congenital malformation of face and neck, unspecified	1	20.0
<b>Circulatory system</b> (n=1)	Congenital absence and hypoplasia of umbilical artery and VSD	1	100.0
<b>Orao facial</b> (n=49)	Other congenital malformations of nose	1	2.0
	Cleft hard palate with cleft soft palate, bilateral	1	2.0
	Cleft hard palate with cleft soft palate, unspecified	12	24.5
	Cleft hard palate with cleft soft palate, unilateral	3	6.1
	Cleft hard palate with cleft soft palate, unspecified	1	2.0
	Central complete cleft palate	1	2.0
	Cleft palate, unspecified, unilateral	1	2.0
	Cleft lip, bilateral	10	20.4
	Cleft lip, specified as unilateral	11	22.5
	Cleft palate with cleft lip	1	2.0
	Cleft hard palate with cleft lip, bilateral	2	4.0
	Cleft hard palate with cleft lip, specified as unilateral	2	4.0
	Cleft hard and soft palate with cleft lip, bilateral	1	2.0
	Cleft hard and soft palate with cleft lip, specified as unilateral	1	2.0
Cleft hard and soft palate with cleft lip NOS	1	2.0	
<b>Digestive system</b> (n=3)	Congenital absence, atresia and stenosis of duodenum	1	33.3
	Congenital absence, atresia and stenosis of rectum without fistula	2	66.6
<b>Congenital malformations of genital organs</b> (n=8)	Congenital rectovaginal fistula	1	12.5
	Congenital malformation of clitoris	2	25.0
	Hypospadias, balanic	3	37.5
	Hypospadias, penile	1	12.5
	Hypospadias, perineal	1	12.5
<b>Congenital malformations and deformations of the musculoskeletal system</b> (n=31)	Talipesquinovarus	11	35.5
	Other specified congenital musculoskeletal deformities	1	3.2
	Accessory finger (s)	2	6.5
	Fused fingers	1	3.2
	Webbed fingers	1	3.2
	Polysyndactyly	1	3.2
	Congenital complete absence of upper limb (s)	2	6.5
	Congenital absence of hand and finger (s)	2	6.5
	Congenital absence of finger (s)	1	3.2
	Congenital absence of foot and toe (s)	2	6.5
	Split foot	1	3.2
	Hypertelorism	1	3.2
	Exomphalos	1	3.2
	Gastroschisis	4	13.0

Nepal also revealed musculoskeletal system as second most common system involved in congenital malformations.<sup>17</sup> The predominance of musculoskeletal and facial malformation could be related to easiness for diagnosis and its visibility during routine physical head to toe examination.

Antenatal visits are an important part of prenatal care. These visits aim at ensuring a normal pregnancy with the delivery of a healthy baby from a healthy mother. They are also the most valuable screening tool to pick up congenital anomalies. Inadequate ANC visits (< 4) have previously been associated with the occurrence of congenital anomalies.<sup>22,23</sup>

Maternal cigarette smoking and alcohol consumption have previously been reported as risk factors for the occurrence of congenital anomalies including oro-facial clefts and congenital heart disease.<sup>24,25</sup> The current study did not show a significant difference in smoking or alcohol consumption during pregnancy when comparing mothers of newborns with or without a diagnosis of congenital malformation.

Female babies had significantly higher congenital anomalies than males in the present study. Socioeconomic status of the included cases was mostly of low socioeconomic status. This could be one of the reasons for a high percentage of affected patients in our setting as socioeconomic status is an important risk factor for birth defect.<sup>25</sup> This may be correlated with the antenatal visits, which is regularly conducted in higher socioeconomic population than in lower socioeconomic population.

Among all congenital anomalies, cardiovascular and central nervous system anomalies led to higher mortality. This could be due to the fact that these systems had direct involvement with life and these babies would mostly require intensive care and interventions, which may not be easily available in most of the resource limited countries. Hence, we also noted high mortality among those with complex congenital heart disease and hydrocephalus with spine-bifida due to unavailability of advanced neonatal cardiac and neuro-surgery service facility in our centre. Our study is a single hospital-based study and hence the results cannot be generalised to the entire country. We also could not check for chromosomal abnormalities and metabolic diseases due to the lack of resources. Hence, we could not comment upon chromosomal anomalies and metabolic disease in this study.

## CONCLUSIONS

The prevalence of congenital malformation in this study was 2.1 per 1000 live births. The most common congenital anomalies involved oro-facial, musculoskeletal system and central nervous system. Lack of routine antenatal visits, lack of folic acid during pre-conception period and low socioeconomic status were the most common risk factors identified for congenital anomalies in our study.

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